Fortnightly

CME DIGEST

REACHING CONTINUING MEDICAL EDUCATION CONVENIENTLY

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Welcome to CME Digest. This fortnightly journal is a new offering from Asian Society of CME, whose vision is to enhance the quality of healthcare service provided through education initiatives. The objective of the journal is to update the knowledge and enhance the skills of physicians in managing both commonly and not so commonly encountered diseases in the clinic. Each issue of the journal would dwell in depth into a disease condition and would be authored by a leading nationally renowned Key Opinion Leader.

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Sincerely

Dr. Sunil Pandey

Director - CME Affairs

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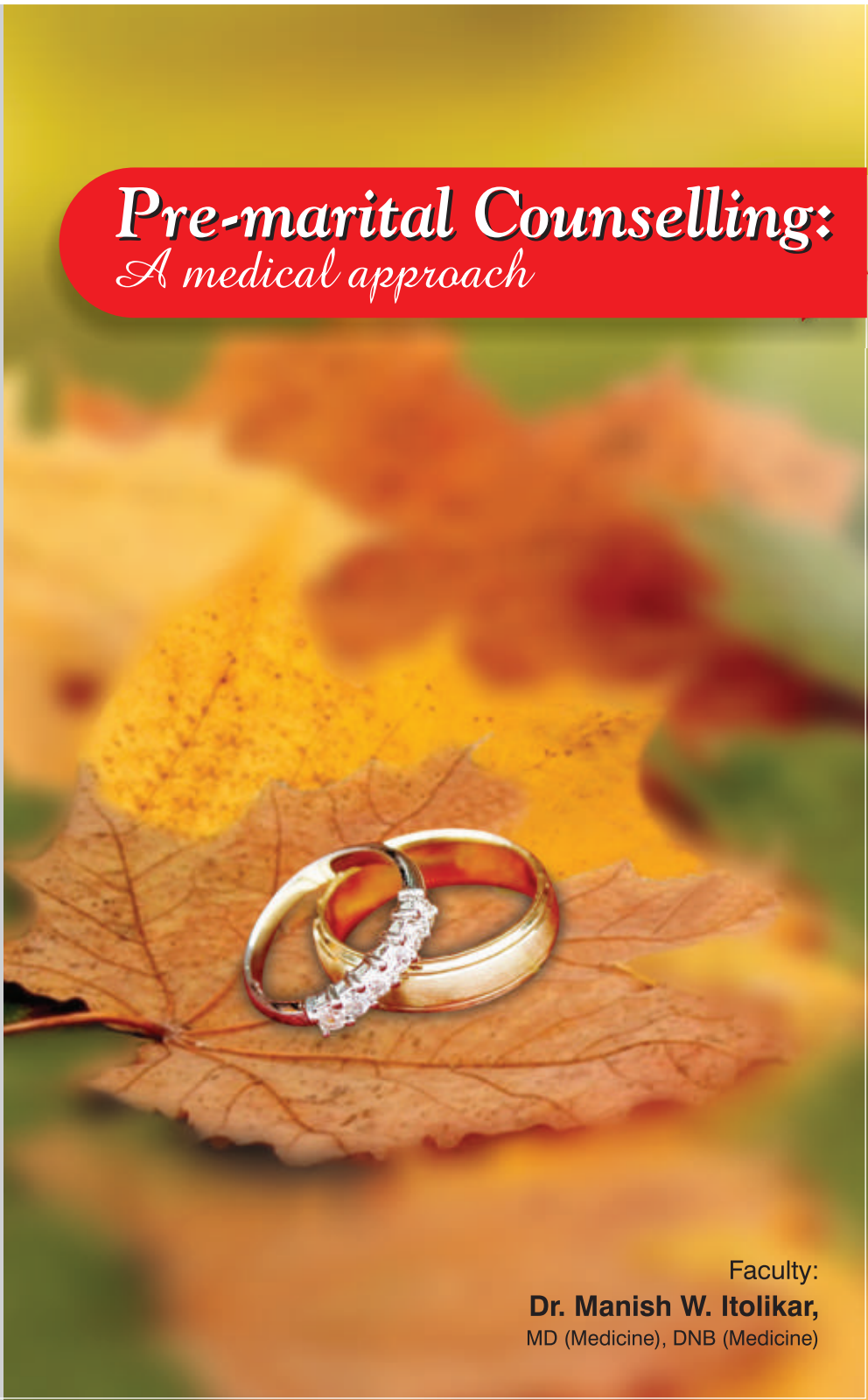
Pre-marital Counselling:
A medical approach



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Editor: Dr. Milind Nadkar **RNI No. MAHENG/2011/37223**

Editorial

Pre-marital counselling: A medical approach

In India, pre-marital counselling is a totally neglected subject. In our country, given the level of socioeconomic status, pre-marital counselling has tremendous importance. There are very few centres for pre-marital counselling and a miniscule number of couples take advantage of these facilities. Spreading the message of importance of pre-marital counselling is the need of the hour. This message needs to be spread not just in public but also in medical fraternity as many medical practitioners are also ignorant about various aspects of counselling dealt with in these centres.

Apart from various communicable diseases and some hereditary conditions that can be passed to the spouse and to the foetus, respectively, these centres also give advise regarding the contraceptive methods to plan the pregnancies. To protect oneself from diseases spread from spouse is ones right, similarly, to protect oneself from hereditary diseases has to be the right of foetus.

It is very important to make couples realise the genetics of X and Y genes to avoid future marital discords. Surely, all of us know about all the aspects discussed in this issue of CME Digest. However, we have to reinforce on the importance of premarital counselling. In future, if one comes across any young 'eligible' boys and girls, please ensure that they are made knowledgeable about pre-marital counselling.

Dr. Milind Nadkar
Editor

Asian Society of Continuing Medical Education

"Asian Society of Continuing Medical Education" is a registered charitable society and not for profit forum of doctors engaged in updating the skills and knowledge of practicing doctors by providing Continuing Medical Education (CME) activities.

Asian Society has worked with many renowned senior faculty in the medical fraternity to create Continuing Medical Education programmes in Live and Home Study formats, leveraging the evidence based knowledge and skills of the thought leaders drawn from various medical specialties and reaching out to a large number of practicing doctors across the country. The Distance Education mode has enabled practicing doctors even from the remotest parts of India to easily and conveniently participate in the programmes without any sacrifice to their practice.

CME Digest is a new and unique initiative from Asian Society of Continuing Medical Education. It is India's first fortnightly Journal dedicated to CME. It will be published from Mumbai under the Editorship of Dr. Milind Nadkar.

The objective of the Journal is to update the knowledge and enhance the skills of physicians in managing both commonly and not so commonly encountered disease conditions in the clinic. Asian Society of Continuing Medical Education through this Journal aims to provide quality CME to doctors in every nook and corner of India.

Each issue of the Journal would dwell in depth on a disease condition and would be authored by a leading nationally renowned Key Opinion Leader.

The CME will be structured for easy and quick assimilation of knowledge. The CME would be supported by Case Studies and Clinical Challenges. The Case Studies would serve the purpose of demonstrating the application of the knowledge while the Clinical Challenges would serve the role of self evaluation.

In addition to the CME, each issue would have columns on News & Notes and Medicolegal.

Editorial Board

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News & Notes

1. Genetic cues to male reproductive birth defects

The causes of cryptorchidism, which is due to the failure of one or both testes to move to the scrotum during foetal development occuring in about 3 percent of full term male births, and hypospadias, which is due to abnormal placement of the opening of the urethra on the penis, occurring in about 1 in 125 births, are mainly unknown. Scientists used comparative genomic hybridisation technique to analyse children with these abnormalities. This technique particulary identifies alterations in chromosomal regions with duplication or deletions that are too small to be observed under a microscope, termed copy number variations. Such alterations can modify gene dosage (gene gains or losses) leading to a change in cell function. They identified that the birth defects in the children were due to variation in the number of copies of *VAMP7* gene. Microduplication on the X chromosome led to a change in oestrogen receptor and androgen receptor action. The interconnection between androgen and oestrogen functioning to control the differentiation of the male reproductive system depends on the balance between androgen and oestrogen action. Gene duplication occurred in 1.35 percent of the 324 subjects, but was absent in approximately 9,000 control subjects who did not have birth defects.

Mouse models that mimicked the human genomic duplication were used to confirm the relationship of gene duplication with the male reproductive tract birth defects. *VAMP7* belongs to soluble N-ethylmaleimide-sensitive factor activating protein receptor (SNARE) family which is a a large protein superfamily comprising of more than 60 members in yeast and mammalian cells, which points to the role of *VAMP7* gene in causing male birth defects. SNARE protein regulates the movement of other proteins in the cell. A slight variation in the amount of this protein considerably affects the oestrogen receptor action, and androgen receptor action to a lesser extent , leading to male reproductive birth defects.

Source – Nature Medicine; 2014

2. Two new genes associated with intellectual disability identified

Researchers at the Centre for Addiction and Mental Health have found two new genes associated with intellectual disability. Two different studies pointed out the presence of two different genes that can play an important role in intellectual stability.

In the first study, microaaray technique was used to map the genes of a large Pakistani family with intermarriages. Amongst the family, five members of the youngest generation were affected with mild-to-moderate intellectual disability. A truncation in the *FBXO31* gene was identified. *FBXO31* gene plays a role in the manner the proteins are processed during development of neurons, specially in the cerebellar cortex.

In the second study, using the same technique, researchers studied the genes of two families with intermarriage, one Austrian and one Pakistani. A disruption in the *METTL23* gene was identified which was associated with mild recessive intellectual disability. The *METTL23* gene takes part in the methylation process which is crucial to brain development and function. Approximately one per cent of children globally are affected by non-syndromic intellectual disability that is marked by an impaired capacity to learn and process new or complex information, eventually causing a decline in cognitive functioning and social adjustment. Genetic defects are a prime reason for intellectual disability, although trauma, infection and external damage to the unborn foetus can also lead to intellectual disability. The above mentioned studies were a part of an ongoing study of affected families in Pakistan, where the cultural tradition of large families and consanguineous marriages among first cousins raises the chances of inherited intellectual disability in the progeny.

The researchers pointed out that though it is easier to find and trace genes in consanguineous families, these genes are definitely not limited to them. A recent study evaluated that 13 percent of intellectual disability cases among people of European descent result when a person inherits two recessive genes. Such type of autosomal recessive gene mutation has been typically not easier to trace, leading to insufficiency of research in this area. Parents of affected children show no symptoms, and the child should inherit one defective copy of the gene from each parent, so that only one in four offspring is likely to be affected. Smaller families, therefore, show a reduced frequency and are less accommodating to this kind of study.

In all, 42 genes associated with non-syndromic recessive forms of intellectual disability have now been found; it has been projected that up to 2,500 genes might be linked with intellectual disability, most of them being recessive.

Source – Human Genetics and Human Molecular Genetics; 2014

CME – Pre test

Pre-marital counselling: A medical approach

1. **Who is at risk for developing a fatal Rhesus (Rh) incompatibility?**
   a. Any Rh+ foetus
   b. Second Rh– foetus of Rh+ mother
   c. Second Rh+ foetus of Rh– mother
   d. Rh– mother
2. **Pre-marital screening is highly beneficial for which of these couple categories?**
   a. Couples going for consanguineous marriage
   b. If either has/ both have a family history of a serious genetic condition
   c. If they are 'carriers' of the same faulty gene
   d. All of the above
3. **How many sickle cell genes should the child inherit to get the disease?**
   a. One
   b. Two – One from each parent
   c. Three – Two from one parent
   d. None of the above
4. **In what case does the child has the sickle cell trait?**
   a. When the genes are inherited from both the parents
   b. When the gene is inherited from one parent
   c. Both a and b
   d. None of the above
5. **Which of these is the most common symptom of sickle cell disease?**
   a. Weakness
   b. Sickle cell crises
   c. Both a and b
   d. None of the above
6. **Mother-foetus Rh blood type incompatibility problems can occur if the mother is _____ and her foetus is _____.**
   a. Rh+; Rh+
   b. Rh+; Rh–
   c. Rh–; Rh+
   d. Rh–; Rh–
7. **Which of the following statements is true of the Rh blood system?**
   a. It was the first blood type system to be discovered
   b. It is more complex genetically than the ABO system
   c. There are 45 Rh blood types
   d. Both b and c
8. **How many major types of thalassaemia exist?**
   a. Two
   b. Three
   c. Four
   d. Five
9. **Mild anaemia is seen in patients with which of these disorders?**
   a. Alpha thalassaemia trait
   b. Beta thalassaemia trait
   c. Both a and b
   d. None
10. **Which of the following may be a sign of a primary syphilis infection?**
    a. Painful sore on the back of the neck
    b. A painless sore in the genital area
    c. A lesion inside the belly button
    d. A painful sore on the genital area

Ans: 1-c; 2-d; 3-b; 4-b; 5-b; 6-c; 7-b; 8-a; 9-c; 10-b

Pre-marital Counselling: A medical approach

Introduction to pre-marital screening

Pre-marital screening can aid in providing protection against diseases that can be transmitted by sexual interactions between partners. Such a screening procedure can reduce the occurrence of common hereditary blood diseases such as thalassaemia and sickle cell disease. It will also protect prospective partners from contracting contagious diseases from each other. The couple can be aware of the possible genetic conditions that their children can inherit, which gives them a choice to prevent it. This could help families with children affected by genetic or chronic infectious diseases to avoid financial, psychological and social burdens.

The services provided to pre-marital couples include:

- Recognising carriers of haemoglobin disorders as well as the diseased individuals
- Providing treatment for individuals with iron deficiency anaemia, mostly women for reinstatement of iron status prior to pregnancy
- Identifying infected individuals early and refers them to specialists
- Providing medical advice and mental support to people intending to get married
- Providing vaccination for hepatitis B to vulnerable individuals

While advocating pre-marital screening, the significance of counselling and examination as a preventive measure has to be communicated. Typical issues pertaining to family planning and reproductive health have to be explained to the couples, such as:

- Anatomical essentials regarding male and female genital organs
- Physiology of menstruation and pregnancy
- Various family planning techniques
- Most frequent preventable disorders such as Rhesus (Rh) incompatibility and Down's syndrome

The procedure of genetic counselling aids in advising patients and relatives who are at risk of an inherited disorder on the:

- Nature as well as outcomes of a disorder
- Chances of developing or transmitting condition
- Ways for management of the disorder
- Family planning methods in order to avoid or ameliorate the condition

The role of genetic counselling not only involves risk assessment, but also helps in comprehending the cause, inheritance pattern and treatment strategy of a condition. Pre-marital counselling has been confirmed to be one of the most significant approaches for prevention of congenital abnormalities, medical and psychosocial marital difficulties. Such counselling has been found to be one of the most significant measure that can decrease the occurrence of genetic conditions, mostly in cases involving old paternal and maternal age.

The intervention in such individuals comprise:

- Treatment of ailments such as various infections
- Alteration of chronic disease medications for reducing teratogenic risks
- Advise on contraception
- Genetic counselling

The World Health Organisation has put forth various measures for the prevention of genetic conditions such as health education and assessment for detection of couples at risk. Millions of children are diagnosed to be affected by congenital conditions every year, and this subsequently leads to many problems in the family and society. Having a healthy baby is enormously important for each couple, particularly for those who have experienced mental or physically retarded child.

Pre-marital history taking and assessment

History

- Consanguinity or any familial disorders
- Medical conditions like diabetes, tuberculosis, hypertension, sexually transmitted infections (STIs)
- Menstrual history
  - Age of menarche
  - Regularity
  - Duration
  - Heaviness of flow
  - Dysmenorrhoea
  - Vaginal discharge
  - Date of last menstruation
- Prior operations
  - Laparotomy
  - Varicocele
  - Hydrocele
  - Hernia
  - Mumps in men
- Immunisation for Rubella
- Family history of hereditary diseases

General examination

- Signs indicating endocrine disturbances
  - Very short stature can signify Turner's syndrome or pituitary dwarfism
  - Very tall stature can indicate gigantism
  - Size of breasts and condition of nipples
  - Unusual obesity, hirsuitism
- Signs of general ailments
  - Cachexia (tuberculosis – chronic renal illness, etc.)
  - Signs of anaemia
  - Skin lesions
  - Cardiovascular conditions
  - Diabetes
  - Hypertension
- Abdominal examination
  - Distribution of pubic hair
  - Abdominal masses
  - Scars
- Assessment of external genital organs

For females:

- Genital ulcers (herpes, syphilitic ulcers, soft sore, granuloma inguinale or lymphogranuloma venereum) or papillomata
- Condition of labia minorae and clitoris

For males:

- Genital ulcers (herpes, syphilitic ulcers, soft sore, granuloma inguinale or lymphogranuloma venereum)
- Urethral discharge
- Varicocele
- Hypospadias or undescended testicles

Pre-marital assessment – Hereditary blood diseases and infectious diseases

Hereditary blood diseases

Sickle cell disease

In sickle cell anaemia, which is an inherited blood condition, the red blood cells become rigid and sickle-shaped instead of the normal flexible round shape (Figure 1). These sickle-shaped cells carry less oxygen for different tissues in the body. They can also obstruct the small blood vessels and eventually disrupt healthy blood flow.

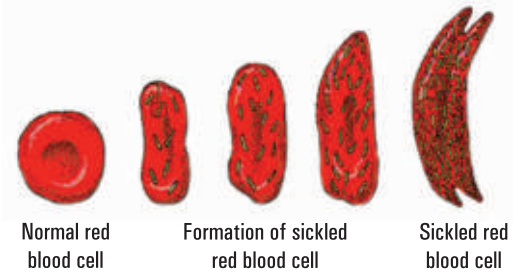


Figure 1: Formation of sickled RBC

Signs and symptoms

The signs and symptoms of sickle cell disease can vary from mild-to-severe and can require frequent hospitalisations due to conditions like:

- Frequent episodes of pain
- Anaemia
  - o Fatigue
  - o Shortness of breath
  - o Pale colour
  - o Loss of appetite
- Jaundice
- Frequent infections
- Hand-foot syndrome that causes swelling of hands and feet
- Splenic atrophy

Inheritance pattern

When both the parents possess the sickle cell trait, i.e., one normal haemoglobin gene and one sickle cell gene, the offspring has (Figure 2):

- 50% probability of inheriting sickle cell trait (one normal gene, one sickle cell gene)
- 25% probability of inheriting sickle cell disease (two sickle cell genes)
- 25% possibility of being at risk of inheriting sickle cell disease (normal)

When one parent possesses sickle cell trait, i.e., one normal gene and one sickle cell gene and the other parent has two normal haemoglobin genes, the offspring has (Figure 3):

- 50% probability of inheriting sickle cell trait (one normal gene and one sickle cell gene)
- 50% possibility of not being at risk of inheriting sickle cell disease (normal)

When one parent has sickle cell disease (two sickle cell genes) and the other parent possesses sickle cell trait (one normal gene, one sickle cell gene), the offspring has (Figure 4):

- 50% probability of inheriting sickle cell trait
- 50% probability of inheriting sickle cell disease

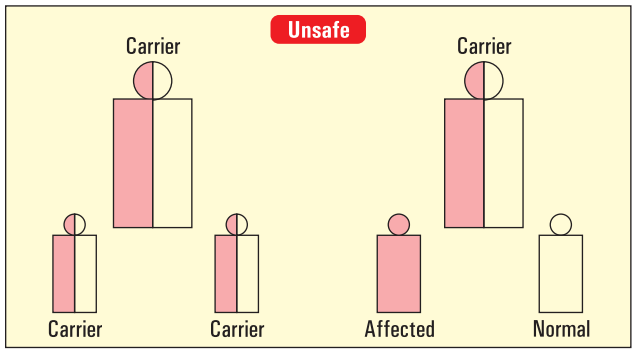


Figure 2: Inheritance pattern when both parents are carriers

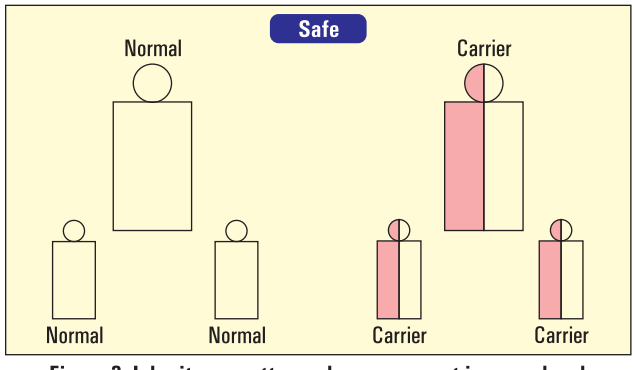


Figure 3: Inheritance pattern when one parent is normal and the other is a carrier

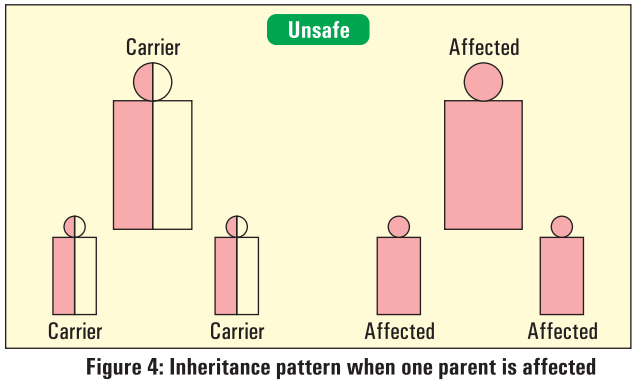


Figure 4: Inheritance pattern when one parent is affected and the other parent is a carrier

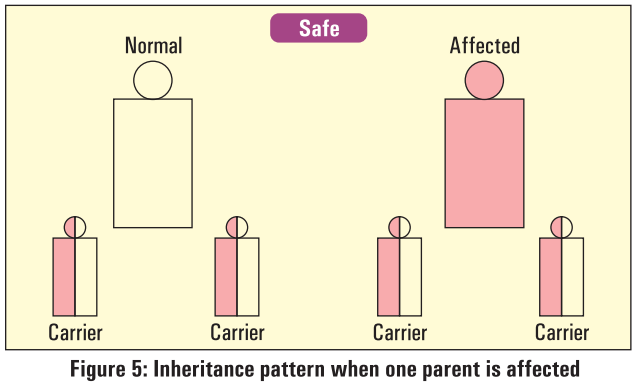


Figure 5: Inheritance pattern when one parent is affected and the other parent is normal

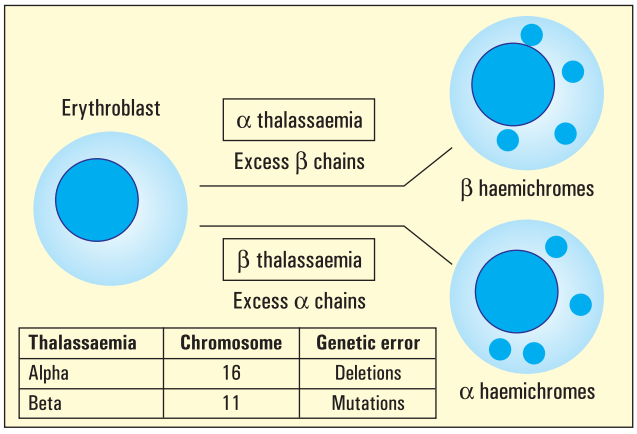
When one parent has sickle cell disease and the other parent possesses two normal haemoglobin genes, the offspring has (Figure 5):

- 100% possibility of inheriting sickle cell trait, but not the disorder

When both parents have sickle cell disease, the child has a 100% possibility of being affected by the disorder.

Thalassaemia

Thalassaemia, which is an inherited blood disorder, leads to generation of irregular haemoglobin concentration. Extreme deterioration of red blood cells leads to anaemia in such patients. The genes coding for haemoglobin are either missing or variant due to thalassaemia. The severe types of thalassaemia are mostly detected in early childhood and are life-long disorders. Alpha and beta thalassaemia are the two primary types of thalassaemia (Figure 6).



Thalassaemia	Chromosome	Genetic error
Alpha	16	Deletions
Beta	11	Mutations

Figure 6: Alpha and beta thalassaemias

Haemoglobin has two types of protein chains known as alpha globin chains and beta globin chains. So, the disorders are called alpha thalassaemia and beta thalassaemia, respectively. Four genes are required in generating the alpha globin section of haemoglobin, two genes from each parent. Alpha thalassaemia is caused when these genes are variant or missing. Two genes influence the generation of the beta globin part of haemoglobin, one from either parent. Thalassaemia can be detected when either one or both of these genes are variants. Thalassaemia minor occurs when one gene is affected and the person is a carrier. The carriers mostly do not present with any signs other than mild anaemia, but their children can inherit the variant genes. Beta thalassaemia majorly occurs when both the genes are variant, and the person eventually has severe anaemia.

Signs and symptoms

The signs of severe anaemia are reported during early childhood that comprises:

- Anaemia
  - o Fatigue
  - o Shortness of breath
  - o Pale colour
  - o Loss of appetite
- Jaundice
- Dark urine
- Protruding abdomen, with enlarged spleen and liver
- Abnormal facial bones and poor growth

Inheritance pattern

When both the parents have been seen to possess the thalassaemia trait, i.e., carriers, the offspring has (Figure 2):

- 50% probability of inheriting thalassaemia trait
- 25% probability of being affected by thalassaemia major
- 25% probability of not being affected by the disease

When one parent is a carrier and the other parent possesses normal haemoglobin genes, the offspring has (Figure 3):

- 50% possibility of having thalassaemia trait
- 50% possibility of not being at risk of inheriting the condition

When one parent has thalassaemia major and the other parent has normal haemoglobin genes, the offspring has (Figure 4):

- 50% possibility of being a carrier
- 50% possibility of presenting with beta thalassaemia

When one parent has thalassaemia major and the other parent has normal haemoglobin genes, the offspring has (Figure 5):

- 100% probability of being affected by the thalassaemia trait, i.e. a carrier, but not the disorder

When both the parents have thalassaemia major, the offspring has:

- 100% probability of inheriting the disorder

Complications

- Osteoporosis
- Gall bladder stones
- Delayed growth
- Dysmorphic features
- Elevated iron concentration in blood that can alter functioning of heart, lungs and body glands

Infectious diseases

Hepatitis B and C

Viral hepatitis is a contagious liver condition, which can deteriorate the functioning of organs. Even though there are 5 major hepatitis viruses, infection due to types B and C lead to chronic disorder, and have been the most frequent reason for liver cirrhosis and cancer. Hepatitis B, which is caused by hepatitis B virus, can be either the acute form of the condition when the disease persists for less than 6 months, or the chronic form of the condition when the disorder lasts for more than 6 months. About 15–25% of the population with chronic hepatitis B eventually develops critical liver problems such as liver damage, cirrhosis, liver failure as well as liver cancer. A series of 3 shots are needed for long-term protection against hepatitis B. This vaccine is 95% efficient in prevention of hepatitis B infection and its adverse outcomes. Hepatitis C, which is caused by hepatitis C virus, does not have any vaccine. 15–25% of people do not need any treatment for clearing the virus, and in 75–85% of people with acute infection it progresses to a chronic form. Roughly 60–70% of individuals with chronic hepatitis C progress to liver disorder, and 1–5% of the people get liver cirrhosis or liver cancer. The apt method for prevention of hepatitis C is by avoiding contamination with infected blood, instruments as well as intravenous drug abuse.

Transmission of hepatitis B and C

- Contact with infected blood
- Sexual contact with an infected person
- Sharing needles, syringes or other injection medication equipment with infected person
- Using an infected individual's personal items contaminated with blood such as razors or toothbrushes
- Tattoo or piercing with unsterilised equipments used on an infected person
- During pregnancy or birth from infected mother to her baby
- Haemodialysis and organ transplant

Signs and symptoms

Numerous people with hepatitis do not present with symptoms. Some of the people can present with mild-to-severe symptoms soon after being infected. Such symptoms include:

- Fatigue and lethargy
- Jaundice
- Fever, headache and joint pain
- Abdominal pain, nausea and vomiting
- Diarrhoea and loss of appetite
- Dark urine and grey-coloured stools
- Skin rash and itching

These symptoms can persist from weeks to various months. The virus can be detected in the blood, even if the individual does not present with any symptom.

Prevention

- Providing vaccination for hepatitis B
- Sterilising surgical equipments and not sharing needles and personnel items
- Ensuring proper screening of blood, plasma, organ and tissue donors
- Refraining from intravenous drug abuse
- Apt testing of pregnant women and born infant (vaccination to be provided)

Syphilis

Syphilis is a sexually transmitted condition, which occurs due to a bacterial infection by *Treponema pallidum* in the genital tract. Syphilis can be transmitted by sexual contact with the infectious lesions, from the mother to foetus *in utero*, during blood product transfusion, and intermittently via breaks in the skin that can be in contact with the infectious lesions. When it is not treated, the condition can progress through 4 phases:

- Primary stage
- Secondary stage
- Latent stage
- Tertiary stage

Transmission

- Sexual contact with an infected individual
- From an infected pregnant woman to her unborn child
- Contact with a syphilis sore that commonly occurs on the external genitals, vagina, anus or in the rectum; however, such sores can be present on the lips and in the mouth
- Sharing needles, syringes or other injection drug tools with infected individual
- Using personal items of an infected person such as razors or toothbrushes

Signs and symptoms

Syphilis is a gradually progressing condition that presents to have various phases. The primary as well as secondary phases of syphilis are highly infectious.

Primary stage

The duration between start of the first symptom after being infected by the bacterium can vary between 3–4 weeks. The main manifestation of this stage is the emergence of a single sore that is called a 'chancre'; however, numerous sores can be present during this stage. This chancre is mostly small, firm, round and painless and appears at the point of contact.

Secondary stage

The secondary stage is typically marked with the development of a rash on one or more regions of the body. This rash mostly does not trigger itching (Figure 7). The rashes due to secondary syphilis can be seen when the chancre is healing or numerous weeks subsequent to the healing of chancre.

Secondary syphilis can also trigger:

- Fever and fatigue
- Swollen lymph glands and sore throat
- Headache

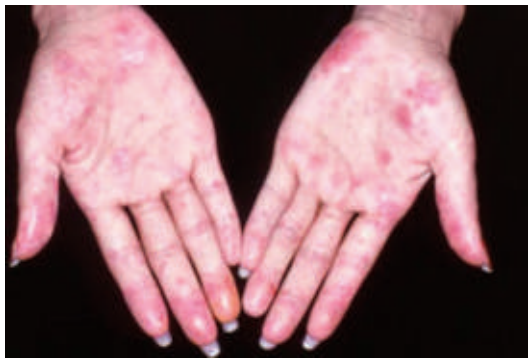


Figure 7: Secondary stage rash on the palms of hands

- Muscle aches
- Patchy hair loss and weight loss

These symptoms mostly resolve within few weeks, however, they can reappear for years.

Tertiary stage

The disorder can possibly deteriorate the functioning of internal organs such as the eyes, heart, liver, bones, brain, nerves and blood vessels. The symptoms associated with this phase are:

- Difficulty in co-ordinating muscle movements
- Paralysis
- Numbness
- Gradual blindness
- Dementia

The damage caused during this phase can lead to death. Syphilis can be transmitted to the baby in the womb when the mother's infection is not treated, which is known as congenital syphilis. The chances of stillbirth or miscarriage are elevated when the baby gets infected.

Human immunodeficiency virus (HIV)/Acquired immunodeficiency syndrome (AIDS)

HIV is a virus that attacks and devastates the infection-fighting cells of the immune system, impairing the system's ability to fight infections. AIDS is the most advanced phase of HIV infection.

Transmission

- Blood transfusion
- Sexual contact with an infected person
- Organ transplant

- Sharing needles, syringes or other injection drug tools with infected person
- Using contaminated personal items of an infected individual
- Infected mother to baby during pregnancy, delivery or breastfeeding

Signs and symptoms

The signs and symptoms of HIV and AIDS differ as per the stage of the infection.

Primary infection

Majority of individuals affected by HIV progress to present with a flu-like condition in 1–2 months subsequent to the entry of virus in the body. This phase can persist for a few weeks.

The associated symptoms are:

- Fever
- Headache
- Muscle aches
- Rash
- Chills
- Sore throat
- Mouth or genital ulcers
- Swollen lymph glands, mostly on the neck
- Joint pain
- Night sweats
- Diarrhoea

The presenting symptoms can be mild and can commonly go unnoticed, but the viral load in the person is mainly elevated during this period. Also, the infection spreads much more rapidly during this stage as compared to the next phase of the infection.

Clinical latent infection

During the clinical latent infection stage, no particular signs and symptoms are seen other than persistent swelling of lymph nodes. This stage persists for 8–10 years. Some people can continue to stay in this phase for a longer period, and others progress to more severe condition comparatively earlier.

Early symptomatic HIV infection

During the early symptomatic infection phase, the virus persists to multiply and devastate the immune cells. Mild infections or chronic symptoms can be seen during this stage such as:

- Fever
- Fatigue
- Swollen lymph nodes – Commonly one of the first signs of HIV infection
- Diarrhoea
- Weight loss
- Cough
- Shortness of breath

Progression to AIDS

The condition progresses to AIDS when no therapy is provided for the HIV infection. The immune system severely deteriorates during this stage, which makes an individual vulnerable to opportunistic infections. The signs and symptoms of these associated symptoms include:

- Soaking night sweats
- Shaking chills or fever higher than 100°F for several weeks
- Cough
- Shortness of breath
- Chronic diarrhoea
- Persistent white spots or unusual lesions on tongue or in mouth
- Headaches
- Persistent, inexplicable fatigue
- Blurred and distorted vision
- Weight loss
- Skin rashes or bumps

Advantages of pre-marital screening

Research has proved that majority of the diseased and carrier individuals are detected by screening the blood. It helps in decreasing the occurrence and transmission of infectious conditions. It also aids in recognition of carriers and infected

individuals at a prior stage, and thereby conducting subsequent treatment and follow-up. The non-immune individuals can be vaccinated. It ultimately protects the society and the future generation from financial, physical and psychological burden due to various communicable conditions.

Necessity of determining ABO and Rh blood group

The clinical importance of blood groups associates with the generation of alloantibodies that travel across the placenta and eventually lead to haemolytic conditions in the foetus and newborn. This depends on:

- Rate of occurrence of alloantigens and alloantibodies
- Functional traits of alloantibodies such as:
  - Thermal range
  - Immunoglobulin class
  - Titre
  - Avidity
  - Ability to fix complement
- Existence of autoantibodies linked to detection and treatment of autoimmune blood conditions

ABO blood group system

The ABO blood group system has been the most significant of all the blood group systems (Figure 8). Four diverse ABO blood groups are present, which aid in ascertaining if the person's red blood cells have the A antigen, the B antigen, both A and B antigens or neither of the antigens. Red blood cell antibodies against the A and B antigens, which are not expressed in the individual's cells, are produced. The antibodies rapidly destroy the red blood cells that possess the antigen.

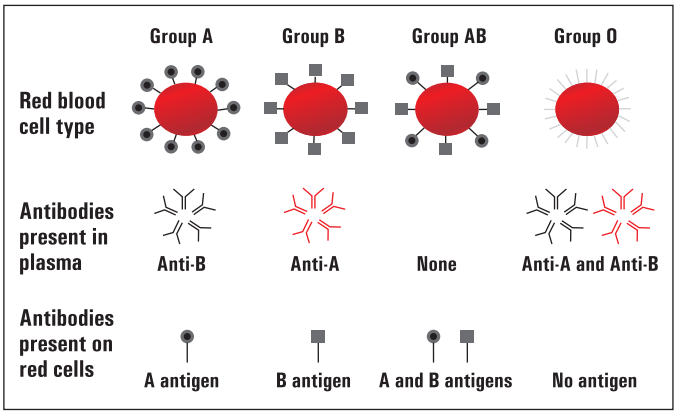


Figure 8: ABO blood groups

ABO inheritance pattern

The ABO blood grouping depends on the ABO gene that is located on chromosome 9. The four blood groups, A, B, AB and O, are differentiated on the basis of inheritance of one or more of the alternative types of this gene or alleles (Table 1).

Table 1: Genetic combination of ABO blood groups

Blood group	Possible genes
A	AA or AO
B	BB or BO
AB	AB
O	OO

The A and B alleles are co-dominant, hence A and B antigens will be expressed on the red cells each time either allele is present. O alleles do not generate A and B antigens, and so are sometimes called 'silent alleles' (Table 2).

Table 2: ABO inheritance patterns

Parental blood groups	Child's blood group
O and O	O
O and A	O or A
O and B	O or B
O and AB	A or B
A and A	A or O
A and B	O or A or B or AB
A and AB	A or B or AB
B and B	O or B
B and AB	B or A or AB
AB and AB	A or B or AB

ABO incompatibility

When a woman with blood group O marries a man with blood group A, B or AB, the foetus' blood group belongs to one of these 3 groups. So, during transplacental haemorrhage, the foetus' blood can get into the mother's circulation and lead to the generation of antibodies in the mother. The blood containing antibodies can get into the circulation of the foetus and eventually destroy the RBCs. Subsequently, this causes neonatal jaundice that appears within 30 hours of birth. Isoimmunisation does not occur when the maternal blood type is either A or B as the naturally produced antibodies are IgM and not IgG. The antibodies are primarily IgG in women with type O blood group, which then leads to haemolysis in the foetus. The involvement of a type A or B foetus with a type O mother has been reported in about 15% of pregnancies. However, haemolytic disease of the newborn occurs in only 3% cases, is severe in only 1%, and <1:1,000 cases need exchange transfusion. The ABO disorder can be seen in first pregnancies, contrasting to the Rh disorder, since anti-A and anti-B antibodies are seen early in life from exposure to A- or B-like antigens present in many foods and bacteria. The clinical presentation of this condition is comparatively much less severe than the Rh disease. The laboratory findings that vary from Rh disease are:

- Smear: Microspherocytosis
- MCV <95, microcytic for a newborn (normal for adult)
- Direct Coombs test is often weakly positive

Rh blood group system

The Rh system incorporates more than 50 antigens that are present on the red cell membrane protein. RhD has been seen to be the most potent and significant antigen. The existence of RhD antigen on the red blood cells provides Rh positivity, and the individuals who do not possess RhD antigen are Rh negative. The exposure of Rh negative people to even smaller levels of Rh positive cells can lead to generation of anti-D alloantibody. The alloantibodies that are directed against the D antigen can possibly cause haemolytic disease of the foetus and newborn.

Rh inheritance pattern

The Rh blood grouping is accountable to two genes, RHD and RHCE that are located on chromosome 1. Assessment of the RhD antigen can aid in ascertaining Rh positivity or Rh negativity. The expression of this RhD antigen varies on the basis of inheritance of the RHD gene from one or both parents. The RHD gene is dominant and so an individual is regarded to be RhD positive when this gene is present, although the gene may have been inherited from one parent. On the contrary, an individual will be RhD negative when the RHD gene is not inherited (Table 3).

Table 3: Rh inheritance patterns

Parental Rh type	Child's Rh type
Positive and Positive	Positive or Negative
Positive and Negative	Positive or Negative
Negative and Negative	Negative

Rh incompatibility

A marriage between Rh+ male and Rh– female is regarded to be biologically incompatible. When both the parents are Rh–, or mother is Rh+ and father is Rh–, or mother is Rh+ and father is Rh+, the marriage is considered to be biologically compatible (Table 4).

Table 4: Rh and marriage compatibility

Boy	Girl	Type of biological marriage
Rh+	Rh+	Compatible marriage
Rh–	Rh–	Compatible marriage
Rh–	Rh+	Compatible marriage
Rh+	Rh–	Incompatible marriage

Haemolytic disease of the newborn

The haemolytic disease of the newborn (HDN), which is also known as erythroblastosis foetalis, isoimmunisation or blood group incompatibility, is seen when foetal red blood cells with an antigen that the mother lacks enter into the maternal circulation. This eventually leads to generation of antibodies that then return to the foetal circulation and trigger RBC destruction.

Pathophysiology

The foetal red blood cells generally move through the placenta towards the maternal circulation during pregnancy. This movement of the foetal RBCs increases significantly

during delivery or termination of pregnancy. The transport of large amount of these RBCs leads to considerable foeto-maternal haemorrhage. In women with Rh– blood carrying a foetus with Rh+ blood, foetal RBCs trigger production of maternal antibody against the Rh antigens. This mechanism remains the same when other antigen systems are involved. No complications are seen during the initial sensitising pregnancy; but, maternal antibodies cross the placenta and lyse foetal RBCs during subsequent pregnancies. This can lead to anaemia, hypoalbuminaemia, and probably high-output heart failure or foetal death. Anaemia prompts the foetal bone marrow to generate and discharge erythroblasts into the foetal peripheral circulation, which is known as erythroblastosis foetalis. Haemolysis causes increased indirect bilirubin concentration in neonates leading to kernicterus. Mostly, isoimmunisation does not produce any symptoms in a pregnant woman.

Clinical presentation

History

Two typical patterns of Rh isoimmunisation severity have been reported. The condition can sustain to be at the same extent of severity or can gradually worsen with each pregnancy. A history of hydropic birth elevates the probability of foetal hydrops in the next pregnancy to 90%. The women at risk for alloimmunisation should generally undertake an indirect Coombs test antibody titres during their initial prenatal visit. The paternal blood type and genotype has to be attained with serological assessment of other Rh antigens.

The markers for severe haemolytic disorder of the newborn comprise:

- Mothers with prior children with haemolytic condition
- Increasing maternal antibody titres
- Elevating amniotic fluid bilirubin level
- Ultrasonographic confirmation of foetal hydrops such as:
  - Ascites
  - Oedema
  - Pleural and pericardial effusions
  - Deterioration of biophysical profile
  - Decreasing haemoglobin levels

Symptoms

Each baby can present with symptoms in a different way. The symptoms during pregnancy include:

- Amniotic fluid can be yellow coloured, with presence of bilirubin
- Ultrasound of foetus shows enlarged liver, spleen or heart and fluid accumulation in the abdomen, around the lungs, or in the scalp of the foetus

The symptoms after birth include:

- Pale colour due to anaemia
- Jaundice or yellow colouring of amniotic fluid, umbilical cord, skin and eyes
- Enlarged liver and spleen
- Hydrops foetalis – Severe oedema of entire body, extreme paleness and difficulty in breathing

Physical examination

An infant born to an alloimmunised mother presents with clinical signs that depend on the severity of the condition. The characteristic diagnostic observations are:

- Jaundice
- Pallor
- Hepatosplenomegaly
- Foetal hydrops in severe cases

Jaundice is mainly noticeable at birth or in the first 24 hours after birth with quickly elevated unconjugated bilirubin concentration. Conjugated hyperbilirubinaemia can be present due to placental or hepatic malfunctioning in infants with severe haemolytic condition. Anaemia commonly occurs because of destruction of antibody-coated RBCs by the reticuloendothelial system. Anaemia can also be caused due to intravascular destruction in certain infants. Extramedullary haematopoiesis can cause hepatosplenomegaly, portal hypertension and ascites. Extreme hepatic extramedullary haematopoiesis leads to portal and umbilical venous hindrance and weakens placental perfusion due to oedema. The placental transport is hindered due to increased placental weight and oedema. Foetal hydrops is a consequence of foetal hypoxia, anaemia, congestive cardiac failure and hypoproteinaemia secondary to hepatic dysfunction.

Hydrops mostly does not occur until the haemoglobin concentration decreases below to about 4 g/dL.

Laboratory findings

The laboratory findings differ with severity of the condition and incorporate:

- Anaemia
- Hyperbilirubinaemia (cord blood bilirubin > 4 mg/dL specifies severe isoimmunisation)
- Reticulocytosis (6–40%)
- Increased nucleated RBC count (>10/100 WBCs)
- Thrombocytopaenia
- Leukopaenia
- + Direct antiglobulin test
- Hypoalbuminaemia
- Rh– blood type
- Smear: Polychromasia, anisocytosis, no sphaerocytes

Treatment

When intrauterine peritoneal transfusion was the only treatment method, newborns were consistently delivered at 32 weeks' gestation. However, this led to an elevated occurrence of hyaline membrane disease and exchange transfusions. After the discovery of intravascular transfusion (IVT) *in utero,* it has been a common treatment approach for a severely affected foetus with delivery planned at term. Lung maturity is not seen frequently in such foetuses due to contamination of amniotic fluid with residual blood during transfusion. However, maternal steroid use can be preferred when delivery is planned before 34 weeks' gestation in order to improve foetal lung maturity. Extreme levels of amniotic fluid bilirubin concentration cause false elevation in the fluorescence depolarisation assays for foetal lung maturity test. Hence, other assessment methods to ascertain foetal lung maturity have to be used such as:

- Infrared spectroscopy
- Lamellar body count
- Phosphatidylglycerol quantitation
- Lecithin/sphingomyelin (L/S) ratio

The intraperitoneal transfusion (IPT) is repeated in cases when the foetal haemoglobin is seen to be lower than 10 g/dL. A second IPT is conducted 10 days subsequent to the first transfusion to increase the haemoglobin level of more 10 g/dL. The next transfusion is conducted every 4 weeks until the time of planned delivery at 34–35 weeks' gestation.

Maternal complications in such cases include:

- Infection
- Transplacental haemorrhage

Foetal complications in such cases are:

- Overtransfusion
- Exsanguination
- Cardiac tamponade
- Infection
- Preterm labour
- Graft versus host disease

The complications during the procedure of IVT have been seen to be:

- Transient foetal bradycardia
- Cord haematoma
- Umbilical vein compression
- Foetal death

Fortunately, IVT provides various benefits like:

- Immediate correction of anaemia
- Resolution of foetal hydrops
- Decreased rate of haemolysis and ensuing hyperinsulinaemia
- Acceleration of foetal growth – For nonhydropic foetuses, IVT has been the only method for moribund hydropic foetuses and those with anterior placenta. Algorithms for management of the first affected pregnancy and the pregnancy in a mother with formerly affected foetus have been provided in Figures 9 and 10. Extensive plasmapheresis with partial replacement using 5% albumin and intravenous immunoglobulin (IVIG) or the administration of IVIG at 1 g/kg body weight weekly have been considered to be reasonably efficient, but these practices can merely delay the need for percutaneous umbilical blood sampling (PUBS) and IVT until 20–22

weeks' gestation. Comparable routine of assessments and treatment are commonly employed in case of pregnancies affected by non-RhD alloimmunisation, such as anti-Rhc, anti-K (K1) and anti-M.

ABO incompatibility

The treatment of hyperbilirubinaemia is highly significant in newborns with ABO incompatibility. The methods for exchange transfusion and phototherapy are similar to that seen in patients with Rh alloimmunisation. The use of IVVIG has been seen to be very efficacious when given during the early phases of the condition. Tin or zinc protoporphyrin should be given intramuscularly at a dose based on body weight.

The probable toxic effects of tin or zinc protoporphyrin include:

- Skin photosensitisation
- Iron deficiency
- Inhibition of carbon monoxide generation

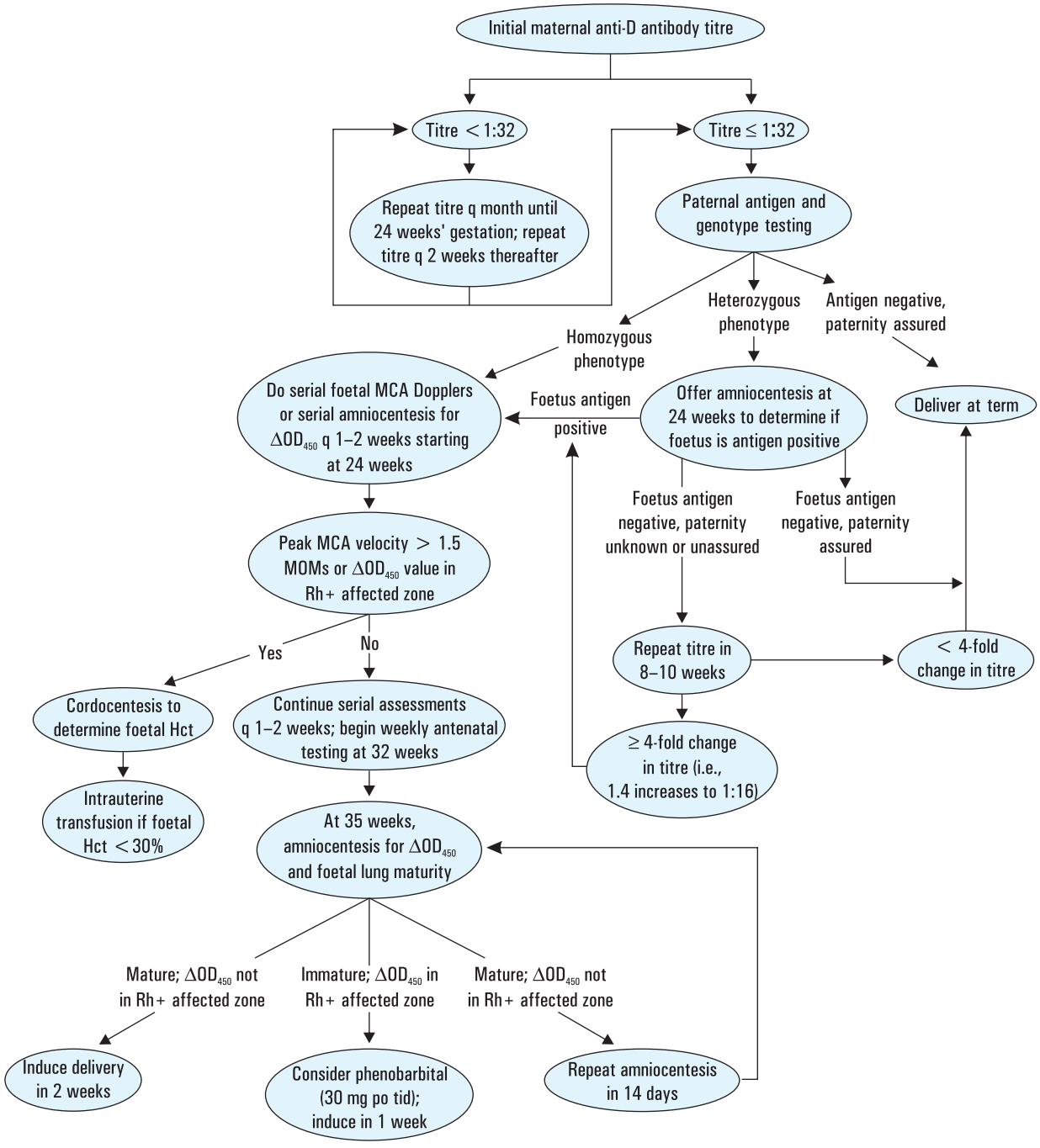


Figure 9: Treatment of first affected pregnancy

Minor blood group incompatibility

Minor blood group incompatibility, which is infrequent, has been seen in approximately 0.8% of pregnant women and mostly with E, c, Kell, Kidd or Duffy antigens. The clinical presentation is similar to the Rhesus disease. The anti-Kell disorder can be severe because of haemolysis or erythroid suppression. Lewis antigen triggers only IgM generation, so maternal antibody screen can be positive, but the foetus remains unaffected.

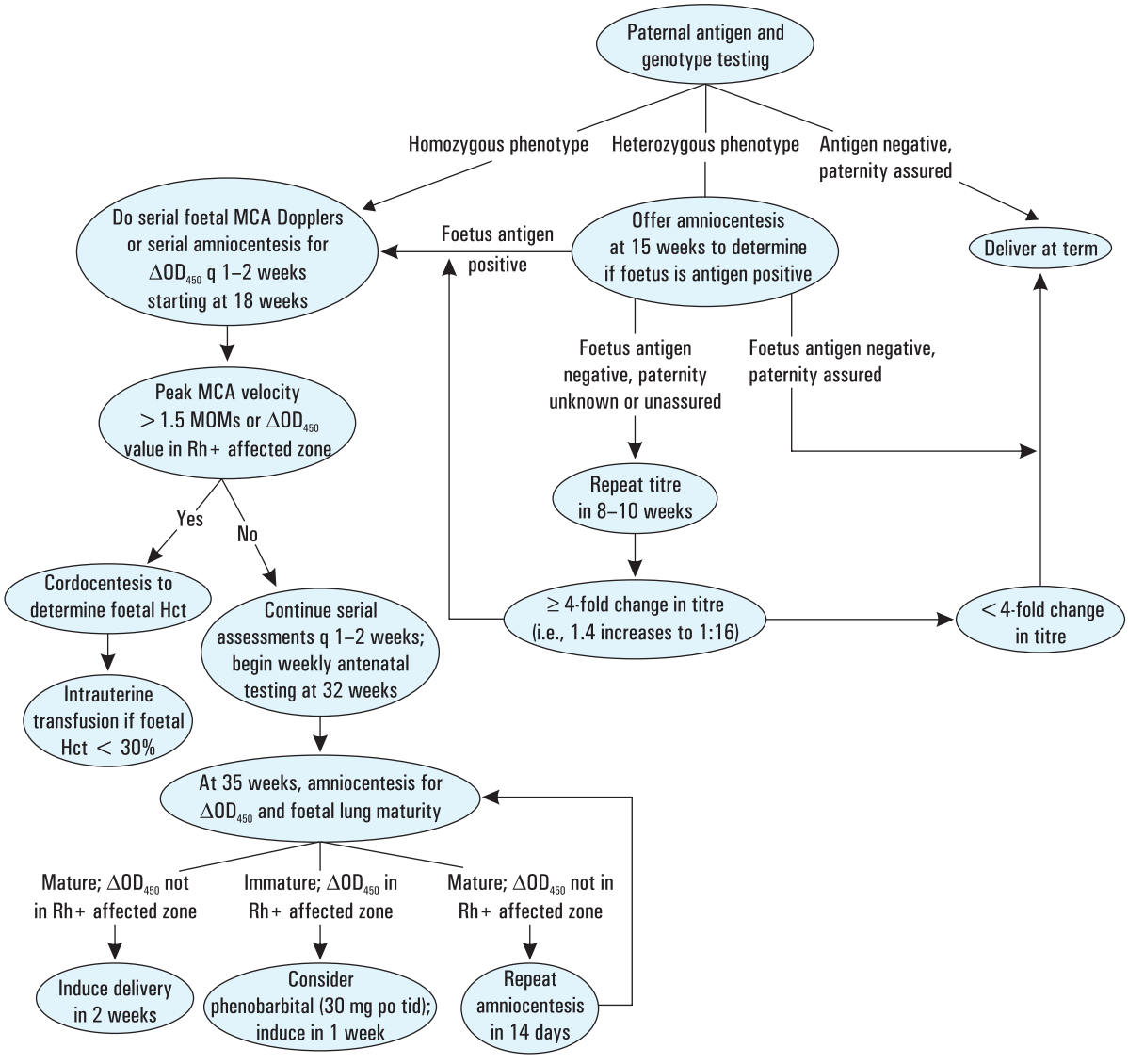


Figure 10: Treatment of pregnant woman with formerly affected foetus

Key insights

- Pre-marital counselling plays a crucial role in preventing various hereditary conditions and infectious diseases.
- Pre-marital counselling aids in identification of carriers and infected individuals at an initial stage.
- Pre-marital counselling helps to protect the society and the future generation from financial, physical and psychological burden due to various communicable conditions.

Birth control methods

There are various methods for birth control and it is very important to choose the most apt technique. Majority of these methods do not provide any protection against sexually transmitted conditions. The effectiveness of a birth control method is estimated by how efficacious it is in preventing pregnancies with typical as well as perfect use.

Birth control pill

The birth control pill, which has to be taken once a day, contains female hormones progestin and oestrogen. The pill prevents the ovary from releasing the egg. It functions by altering the lining of the uterus and makes it difficult for the egg to attach to the uterus wall. Also, it thickens the cervical mucous and makes it hard for the sperm to pass into the uterus.

Advantages

- Effectiveness: 92 – 99.7%
- Continuous protection when taken appropriately
- Easily reversible, non-contraceptive health benefits
- Menstrual period can be lesser in flow and shorter in duration, and causes less cramping
- Can decrease risk of certain cancers of reproductive system
- Can improve acne

Disadvantages

- No protection against STIs, including HIV
- Have to be taken about the same time each day to maintain a balanced level of hormones in the body
- Certain side effects with birth control medications include:
  - o Stomach upset
  - o Breast tenderness
  - o Bleeding between periods
  - o Headaches
- Women with following conditions may not be able to take the pills due to elevated risk of blood clot:
  - o Smokers over 35 years of age
  - o High blood pressure

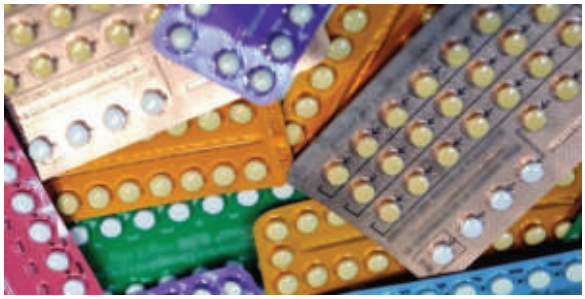


Figure 11: Birth control pill

Transdermal patch

The transdermal patch, which contains polyester, comprises of the female hormones oestrogen and progestin. It functions similarly as the birth control medications, such as:

- Stopping ovary from releasing the egg
- Altering lining of the uterus and making it difficult for the egg to attach to the uterus wall
- Thickening cervical mucus and makes it hard for the sperm to pass into the uterus

A new patch has to be used every week for a period of 3 weeks.

Advantages

- Effectiveness: 92 – 99.7%
- Used weekly, rapidly reversible, continuous protection when properly used
- Continuous protection when taken appropriately
- Easily reversible, non-contraceptive health benefits

Figure 12: Transdermal patch

Note: Contd on page 25

QUICK REFERENCE PULL OUT

Detach & file for future reference

Introduction to pre-marital screening

Pre-marital screening can aid in providing protection against diseases that can be transmitted by sexual interactions between partners. Such a screening procedure can reduce the occurrences of common hereditary blood diseases such as thalassaemia and sickle cell disease. It will also protect prospective partners from contracting contagious diseases from each other. The couple can be aware of the possible genetic conditions that their children can inherit, which gives them a choice to prevent it. This could help families with children affected by genetic or chronic infectious diseases to avoid financial, psychological and social burdens. While advocating pre-marital screening, the significance of counselling and examination as a preventive measure has to be communicated. The typical issues pertaining to family planning and reproductive health have to be explained to the couples, such as:

- Anatomical essentials regarding male and female genital organs
- Physiology of menstruation and pregnancy
- Various family techniques
- Most frequent preventable disorders such as Rhesus incompatibility and Down's syndrome

The role of genetic counselling not only involves risk assessment, but also helps in comprehending the cause, inheritance pattern and treatment strategy of a condition. Pre-marital counselling has been confirmed to be one of the most significant approaches for prevention of congenital abnormalities, medical and psychosocial marital difficulties. Such counselling is one of the most significant measure that can decrease the occurrence of genetic conditions, mostly in cases involving old paternal and maternal age.

The World Health Organisation has put forth various measures for the prevention of genetic conditions such as health education, assessment for detection of couples at risk. Millions of children are diagnosed to be affected by congenital conditions every year, and this subsequently leads to many problems in the family and society. Having a healthy baby is essentially important for each couple, particularly for those who have experienced mental or physically retarded child.

Premarital assessment: Hereditary blood diseases and infectious diseases

Hereditary blood diseases

Sickle cell disease

In sickle cell anaemia, which is an inherited blood condition, the red blood cell becomes rigid and sickle-shaped instead of the normal flexible round shape. These sickle-shaped cells carry less oxygen to various tissues in the body. They can also obstruct the small blood vessels and eventually disrupt healthy blood flow.

Signs and symptoms

The signs and symptoms of sickle cell disease can vary from mild-to-severe and can require frequent hospitalisations for conditions like:

- Frequent episodes of pain
  - Anaemia
  - Fatigue
  - Shortness of breath
  - Pale colour
  - Loss of appetite
- Jaundice
- Frequent infections
- Hand-foot syndrome that causes swelling of hands and feet
- Splenic atrophy

Thalassaemia

Thalassaemia, which is an inherited blood disorder, leads to generation of irregular haemoglobin concentration. Extreme deterioration of red blood cells leads to anaemia in such patients. The genes coding for haemoglobin are either missing or variant due to thalassaemia. Severe types of thalassaemia are mostly detected in early childhood and are life-long disorders. Alpha and beta thalassaemia are the two primary types of thalassaemia.

The signs of severe anaemia are reported during early childhood that comprises:

- Anaemia
  - Fatigue
  - Shortness of breath
  - Pale colour
  - Loss of appetite
- Jaundice
- Dark urine

- Protruding abdomen, with enlarged spleen and liver
- Abnormal facial bones and poor growth

Inheritance pattern

When both the parents have been seen to possess the thalassaemia trait, i.e., carriers, the offspring has:

- 50% probability of inheriting thalassaemia trait
- 25% probability of being affected by thalassaemia major
- 25% probability of not being affected by the disease

When one parent is a carrier and the other parent possesses normal haemoglobin genes, the offspring has:

- 50% possibility of having thalassaemia trait
- 50% possibility of not being at risk of inheriting the condition

When one parent has thalassaemia major and the other parent has normal haemoglobin genes, the offspring has:

- 50% possibility of being a carrier
- 50% possibility of presenting with beta thalassaemia disease

When one parent has thalassaemia major and the other parent has normal haemoglobin genes, the offspring has:

- 100% probability of being affected by the thalassaemia trait, i.e. a carrier, but not the disorder

When both the parents have thalassaemia major, the offspring has:

- 100% probability of inheriting the disorder

Complications

- Osteoporosis
- Gall bladder stones
- Delayed growth
- Dysmorphic features
- Elevated iron concentration in blood that can alter functioning of heart, lungs and body glands

Infectious diseases

Hepatitis B and C

Viral hepatitis, which is a contagious liver condition, can deteriorate the functioning of the organs. Even though there are 5 major hepatitis viruses, infection due to types B and C lead to chronic disorder, and have been the most frequent reason for liver cirrhosis and cancer. Hepatitis B, which is caused by hepatitis B virus, can be either the acute form of the condition when the disease persists for less than 6 months, or the chronic form of the condition when the disorder lasts for more than 6 months. About 15–25% of the population with chronic hepatitis B eventually develop critical liver problems such as liver damage, cirrhosis, liver failure as well as liver cancer. A series of 3 shots are needed for long-term protection against hepatitis B.

Transmission of hepatitis B and C

- Contact with infected blood
- Sexual contact with an infected person
- Sharing needles, syringes or other injection medication equipment with infected person
- Using an infected individual's personal items contaminated with blood such as, razors or toothbrushes
- Tattoo or piercing with unsterilised equipments used on an infected person
- During pregnancy or birth from infected mother to her baby
- Haemodialysis and organ transplant

Signs and symptoms

Numerous people with hepatitis do not present with symptoms. Some of the people can present with mild-to-severe symptoms soon after being infected. Such symptoms include fatigue, lethargy, jaundice, fever, headache, joint pain, abdominal pain, nausea, vomiting, diarrhoea, loss of appetite, dark urine, grey-coloured stools, skin rash and itching. These symptoms can persist from weeks to various months. The virus can be detected in the blood, even if the individual does not present with any symptom.

Prevention

Providing vaccination for hepatitis B

Syphilis

Syphilis, which is a sexually transmitted condition, occurs due to a bacterial infection by *Treponema pallidum* in the genital tract. Syphilis can be transmitted by sexual contact with the infectious lesions, from the mother to foetus *in utero,* during blood product transfusion, and intermittently via breaks in the skin that can be in contact with the infectious lesions. When it is not treated, the condition can progress through 4 phases:

- Primary stage
- Secondary stage
- Latent stage
- Tertiary stage

Transmission

- Sexual contact with an infected individual
- From an infected pregnant woman to her unborn child
- Contact with a syphilis sore that commonly occurs on the external genitals, vagina, anus or in the rectum; however, such sores can also be present on the lips and in the mouth
- Sharing needles, syringes or other injection drug tools with infected individual
- Using personal items of an infected person such as razors or toothbrushes

Signs and symptoms

Syphilis is a gradually progressing condition that presents to have various phases. The primary as well as secondary phases of syphilis are highly infectious.

1. Primary stage

The duration between start of the first symptom after being infected by the bacterium can vary between 3–4 weeks. The main manifestation of this stage is the emergence of a single sore that is called chancre; however, numerous sores can be present during this stage. This chancre is mostly small, firm, round and painless

2. Secondary stage

The secondary stage is typically marked by occurence of rash on one or more regions of the body. This rash mostly does not trigger itching. The rashes due to secondary syphilis can be seen when the chancre is healing or numerous weeks subsequent to the healing of chancre.

Secondary syphilis can also trigger fever and fatigue, swollen lymph glands and sore throat, headache, muscle aches, patchy hair loss and weight loss. These symptoms will mostly resolve within a few weeks, however, they can reappear for years.

3. Tertiary stage

The disorder can possibly deteriorate the functioning of the internal organs such as the eyes, heart, liver, bones, brain, nerves and blood vessels. The symptoms associated with this phase are:

- Difficulty in co-ordinating muscle movements
- Paralysis
- Numbness
- Gradual blindness
- Dementia

The damages during this phase can lead to death. Syphilis can be transmitted to the baby in the womb when the mother's infection is not treated, which is known as 'congenital syphilis'. The chances of stillbirth or miscarriage are elevated when the baby gets infected.

Prevention

- Sterilising surgical equipments and not sharing needles and personnel items
- Ensuring proper screening of blood, plasma, organ and tissue donors
- Refraining from intravenous drug abuse
- Apt testing of pregnant women and born infant (vaccination to be provided)

Advantages of pre-marital screening

Research has proved that majority of the diseased and carrier individuals have been detected by screening the blood. It helps in decreasing the occurrence and transmission of infectious conditions. It also aids in recognition of carriers and infected individuals at a prior stage, and thereby conduct subsequent treatment and follow-up. The non-immune individuals can be vaccinated. It ultimately protects the society and the future generation from financial, physical and psychological burden due to various communicable conditions.

Birth control methods

There are various methods for birth control and it is very important to choose the most apt technique. Majority of these methods do not provide any protection against sexually transmitted conditions.

Birth control pill

The birth control pill, which has to be taken once a day, contains female hormones progestin and oestrogen. The pill prevents the ovary from releasing the egg. It functions by altering the lining of the uterus and makes it difficult for the egg to attach to the uterus wall.

Advantages

- Effectiveness: 92–99.7%
- Continuous protection when taken appropriately
- Easily reversible, non-contraceptive health benefits
- Menstrual period can be lesser in flow and shorter in duration, and causes less cramping
- Can decrease risk of certain cancers of reproductive system
- Can improve acne

Disadvantages

- No protection against sexually transmitted infections, including HIV

- Has to be taken about the same time each day to maintain a balanced level of hormones in the body

Certain side effects with birth control medications include:

- Stomach upset
- Breast tenderness
- Bleeding between periods
- Headaches

Women with following conditions may not be able to take the pills due to elevated risk of blood clot:

- Smokers over age of 35 years
- High blood pressure

Emergency contraceptive pill

The emergency contraceptive pill either has 2 pills consisting of progestin, or 2 pills consisting of progestin and oestrogen. The functions of this pill include:

- Prevention of ovary from releasing egg
- Prevention of sperm and egg from uniting
- Prevention of fertilised egg from attaching to the wall of the uterus

This pill has to be consumed within 72 hours of unprotected intercourse for preventing pregnancy. Both the pills have to be taken at once, or the first pill as soon as possible and the second pill after 12 hours.

Advantages

- 95% effectual if consumed within 24 hours, 85% effectual if consumed within 25–48 hours, or 58% effectual if consumed within 49–72 hours of unprotected vaginal sex
- Will not cause an abortion or harm the fetus if already pregnant

Disadvantages

- Has to be consumed within 72 hours of unprotected vaginal intercourse
- Can trigger side effects like nausea, mild stomach upset, tiredness, headache or spotting

Diaphragm

A diaphragm is a soft rubber dome that is placed over the cervix and prevents sperm from reaching the cervix. It has to be used with a spermicide, and can be inserted into the vagina up to 6 hours prior to intercourse. It has to be kept inserted for not less than 6 hours subsequent to intercourse, but should not be kept inserted for more than 24 hours due to risk of toxic shock syndrome.

Advantages

- Effectiveness: 84–94%
- Reusable and has to be inserted only when required
- Non-hormonal technique

Disadvantages

- Has to be reinstalled with weight gain/loss of 10 lbs
- Women have to be comfortable with insertion and removal
- Cannot be used during menstrual period
- Elevated chances of bladder and yeast infections and bacterial vaginosis
- Has to be washed after every use and then stored in a cool, dry place
- Can lead to irritation from spermicide or latex in diaphragm, which rises risk of HIV or sexually transmitted diseases when exposed
- Can get displaced

Male condom

Male condom is a thin sheath that is generally made of latex. It prevents the sperm from meeting the egg. It can be combined with other birth control techniques for enhancing effectiveness.

Advantages

- Effectiveness: 85–98%
- Easily obtained and reasonably priced
- Provides protection against certain sexually transmitted diseases, including HIV

Disadvantages

- Can break when not used properly
- New condom has to be used with every act of intercourse

Female condom

The female condom is a soft thin polyurethane sheath that loosely lines the vagina and covers the area outside to prevent the sperm from entering the vagina.

Advantages

- Effectiveness: 79–95%
- Easily available and can be used in women with latex allergies
- Provides protection against STIs, including HIV

Disadvantages

- User has to be comfortable with insertion
- Has to be inserted properly prior to sexual intercourse

Contd. from page 20

- Menstrual period can be lesser in flow and shorter in duration, and causes less cramping
- Can decrease risk of certain cancers of reproductive system
- Can improve acne

Disadvantages

- No protection against STIs, including HIV
- Can be less efficacious in women weighing more than 90 kg
- Can trigger skin irritation at the location where patch is placed
- Certain side effects with the patch include:
  - o Stomach upset
  - o Breast tenderness
  - o Bleeding between periods
  - o Headaches
- Women with following conditions may not be able to take the pills due to elevated risk of blood clot:
  - o Smokers over 35 years of age
  - o High blood pressure

Contraceptive ring

The contraceptive ring is placed into the vagina which releases the female hormones oestrogen and progestin. This stays in the vagina for 21 days, and then removed for 7 days for regular menstruation. It provides the same functions as a contraceptive pill or a patch.

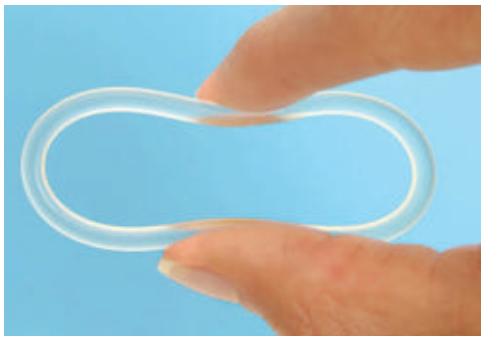


Figure 13: Contraceptive ring

Advantages

- Effectiveness: 92 – 99.7%
- Controls menstruation
- Rarely felt by the woman or partner
- Quickly reversible
- Not consumed and so does not trigger stomach problems

Disadvantages

- Can accidentally fall out under some conditions
- Side effects include:
  - o Nausea
  - o Breast tenderness
  - o Alteration in menses
- Cardiovascular risk in smokers greater than 35 years of age
- Necessitates comfort with self insertion

Injectable progestin

The injection contains the hormone progestin. It has to be administered every 12 weeks. It stops the process of ovulation, thickens the cervical mucous and makes it difficult for the sperm to travel into the uterus.

Advantages

- Effectiveness: 97–99.7%
- Efficient immediately when given within the first 5 days of a normal period
- Can be taken by women who cannot use oestrogen
- Reversible method

Disadvantages

- Delayed return to fertility – Not recommended if planning conception in subsequent 1–2 years

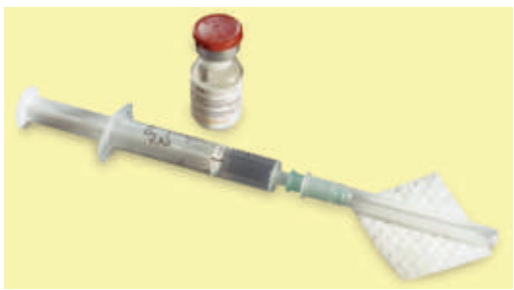


Figure 14: Injectable progestin

- Irregular or absent menses
- Weight gain
- Adverse effects can persist up to 6–8 months after last injection
- Alters bone density – Not recommended for long-term usage

Intrauterine device

Intrauterine device is a small T-shaped device that is placed into the uterus. When this device is hormonal, it contains progestin that is gradually discharged into the bloodstream over a period of 5 years. The non-hormonal device aids in preventing the egg from meeting the sperm and attaching to the wall of the uterus.

Advantages

- Effectiveness: 99.2–99.8%
- Fertility returns to normal after removal
- Nothing to be remembered after insertion
- Can be used by women who cannot tolerate oestrogen
- Hormonal – Can decrease amount of bleeding during periods, so useful for women with heavier periods
- Non-hormonal – Can be effective in women who are unable to tolerate hormonal contraception; can be used safely during breastfeeding, and can be used 6 weeks after giving birth

Disadvantages

- Can trigger irregular menstruation and cramping
- Can cause a rare risk of uterine perforation or pelvic infection
- Strings have to be checked to see if the device is in place
- Increases probability of pelvic inflammatory disease

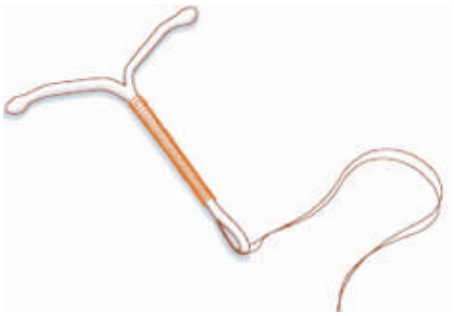


Figure 15: Intrauterine device

Emergency contraceptive pill

The emergency contraceptive pill either has two pills consisting of progestin or two pills consisting of progestin and oestrogen. The functions of this pill include:

- Prevention of ovary from releasing egg
- Prevention of sperm and egg from uniting
- Prevention of fertilised egg from attaching to the wall of the uterus

This pill has to be consumed within 72 hours of unprotected intercourse for preventing pregnancy. Both the pills have to be taken at once, or the first pill as soon as possible and the second pill after 12 hours.

Advantages

- 95% effectual if consumed within 24 hours, 85% effectual if consumed within 25–48 hours, or 58% effectual if consumed within 49–72 hours of unprotected vaginal sex
- Will not cause an abortion or harm the foetus if already pregnant

Disadvantages

- Has to be consumed within 72 hours of unprotected vaginal intercourse
- Can trigger side effects like nausea, mild stomach upset, tiredness, headache or spotting

Figure 16: Emergency contraceptive pill

Diaphragm

A diaphragm is a soft rubber dome that is placed over the cervix and prevents sperm from reaching the cervix. It has to be used with a spermicide, and can be inserted into the vagina up to 6 hours prior to intercourse. It has to be kept inserted for not less than 6 hours subsequent to intercourse, but should not be kept inserted for more than 24 hours due to risk of toxic shock syndrome.

Figure 17: Diaphragm

Advantages

- Effectiveness: 84–94%
- Reusable and has to be inserted only when required
- Non-hormonal technique

Disadvantages

- Has to be reinstalled with weight gain/loss of 10 lbs
- The user has to be comfortable with insertion and removal
- Cannot be used during menstrual period
- Elevated chances of bladder and yeast infections and bacterial vaginosis
- Has to be washed after every use and then stored in a cool, dry place
- Can lead to irritation from spermicide or latex in diaphragm, which rises risk of HIV or STIs when exposed
- Can get displaced

Male condom

Male condom is a thin sheath that is generally made of latex. It prevents the sperm from meeting the egg. It can be combined with other birth control techniques for enhancing effectiveness.

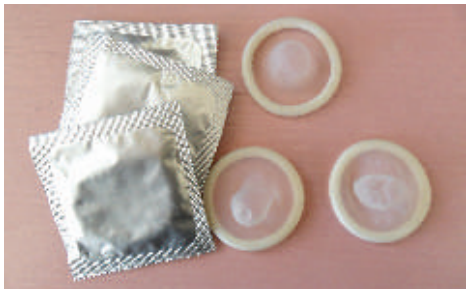


Figure 18: Male condom

Advantages

- Effectiveness: 85–98%
- Easily obtained and reasonably priced
- Provides protection against certain STIs, including HIV

Disadvantages

- Can break when not used properly
- New condom has to be used with every act of intercourse

Female condom

The female condom is a soft thin polyurethane sheath that loosely lines the vagina and covers the area outside to prevent the sperm from entering the vagina.

Advantages

- Effectiveness: 79–95%
- Easily available and can be used in women with latex allergies
- Provides protection against STIs, including HIV

Disadvantages

- User has to be comfortable with insertion
- Has to be inserted properly prior to sexual intercourse

Figure 19: Female condom

Spermicides

Spermicides are chemicals that kill the sperms. They can be used alone or in conjunction with other birth control techniques to elevate effectiveness.

Advantages

- Effectiveness: 71–82% when used alone
- Has to be used when required

Disadvantages

- Do not provide protection against STIs, including HIV

- Can trigger skin irritation, leading to easy transmission of an STI or HIV, if exposed

Figure 20: Spermicides

Sponges

Small, soft foam sponge can be placed into the vagina to cover the cervix. It functions by stopping the sperms from reaching the cervix. It should not be kept in the vagina for more than 30 hours, but it has to be placed for not less than 6 hours subsequent to last intercourse.

Advantages

- Effectiveness: 68–91%
- Non-latex product, so will not trigger allergies

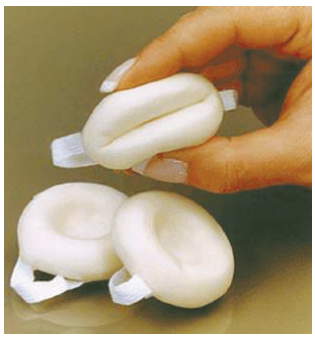


Figure 21: Sponges

- Can be used for more than one act of intercourse
- Easily available

Disadvantages

- Increases risk of being affected by bladder and yeast infections and bacterial vaginosis
- Sponges can trigger some irritation, increasing risk of HIV or STI, if exposed
- Can cause toxic shock syndrome if not removed for more than 30 hours
- Cannot be used during menstrual period
- Person has to be comfortable with insertion and removal

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Case study 1

Case presentation

A 13-year-old male child was brought by his mother to the paediatric emergency department for complaints of abdominal pain and jaundice.

Medical history

- He reported history of mild chronic haemolytic anaemia, elevated bilirubin and cholelithiasis
- At the age of 5 years, he had been reported with an acute haemolytic crisis with splenomegaly
- No other complications associated with sickle cell disease, including painful crisis, pneumonia, acute chest syndrome, or priapism were reported by the patient

On examination

- Height: 5 feet
- Weight: 38 kg
- Temperature: 97.1°F
- RR: 18 breaths/min
- Pulse: 74 beats/min
- Spleen examination: Enlarged, tender
- CT-scan: Spleen size of 21 cm with areas of infarct

Laboratory investigations

- Complete blood count results for the patient and his parents were as shown below in Table 1

Table 1: Complete blood count results

CBC test	Patient	Mother	Father
Erythrocytes (10^6/µL)	3.49	4.10	4.35
Haematocrit (%)	29.3	36.8	38.8
Haemoglobin (g/dL)	10.1	12.1	14.2
Leukocytes (10^3/µL)	5.3	5.7	4.4
Platelets (10^3/µL)	383	254	197
MCV (fL)	84.7	90.7	84.7

MCV: Mean corpuscular volume

- Haemoglobin HPLC test showed results as shown below in Table 2

Table 2: Results for haemoglobin hplc test

CBC test	Patient	Mother	Father
Hb A2, percentage	3.7	2.7	3.6
Hb F, percentage	0.9*	0.3	0.0
Hb S, percentage	51.8	0	37.3
Hb A, percentage	43.1	96.6	58.1

*The patient's low Hb F level may have contributed to the severity of his phenotype
The father is heterozygous for a common α-thalassaemia

- Blood microscopy: Sickle-shaped cell blood cells (Figure 1)

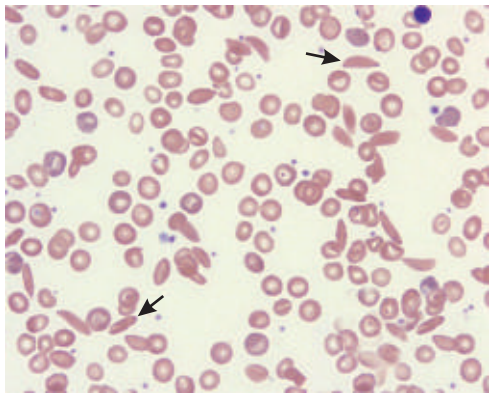

Figure 1: Peripheral blood smear showing sickle-shaped cells

Other investigations

Uniparental disomy (UPD) analysis on chromosome 11: Normal biparental inheritance from 11q25 to 11p13 was reported, whereas from 11p14.2 to 11p15.5 (near the p telomere) the patient had reported decreased levels of the maternal allele. These findings showed mosaic segmental paternal isodisomy that extended from 11p13-11p14.2 to the 11p telomere (Figure 2).

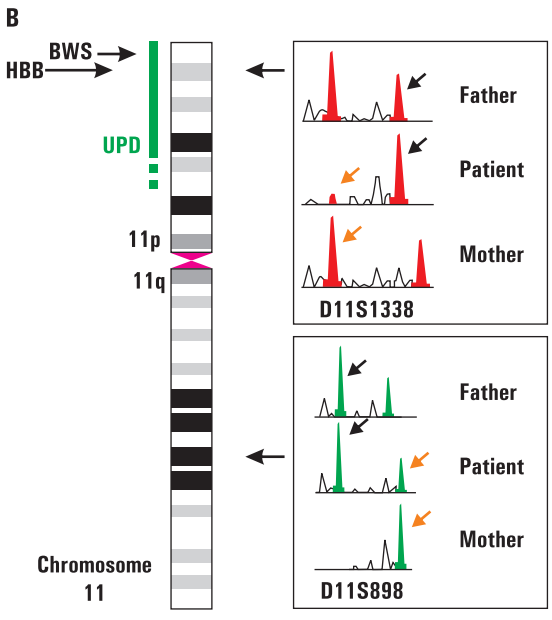

Figure 2: Genotyping results for the patient and his parents

Diagnosis: Mild SCD resulting from postzygotic mitotic recombination leading to uniparental disomy

Treatment

The patient was considered for treatment with IV fluids and IV ceftriaxone for 2 weeks.

Outcome of the treatment

- On follow-up, he reported to have responded well to the treatment
- The patient was advised for regular follow-up, thereafter

Take home message

- Sickle cell disease is a disorder which is presented with recessive Mendelian inheritance, where each parent contributes one mutant allele to an affected offspring
- It is commonly due to homozygosity for the haemoglobin S (Hb S) mutation in the β-globin gene, HBB (on chromosome 11p15.4) that substitutes valine for glutamic acid at codon 6
- Clinical manifestation associated with sickle cell disease usually includes painful crises and splenic sequestration crises

Case study 2

Case presentation

A 26-year-old male patient came to the doctor's clinic for routine check-up. He also had plans to marry the following year, and hence considered this visit as check-up for any blood related infection.

Medical history

- He was reported with asthma, and was on reliever medications
- He was reported to inject drugs during parties with his friend 3 years back, which he had now stopped

Family and personal history

- His father had been diagnosed with hypertension since age of 54 years, and was undergoing antihypertensive treatment
- His mother and elder brother were reported to be healthy
- No other family members were reported with any major illnesses
- He was reported to be an occasional drinker

On examination

- Height: 5 feet 8 inches
- Weight: 67 kg
- Temperature: 101.1°F
- RR: 17 breaths/min
- Pulse: 71 beats/min
- Systemic examination: Unremarkable

Laboratory examinations

- Hb: 12.1 g/dL
- Serum bilirubin: 10.9 μmol/L
- Serum albumin: 47.8 g/L
- International normalized ratio (INR): 1.2
- Aspartate aminotransferase (AST): 80 IU/L
- Alanine aminotransferase (ALT): 158.8 IU/L

- Serum alkaline phosphatase levels: Normal
- Hepatitis C antibody test: Positive
- Recombinant immunoblot assay: Positive
- HIV infection: Negative

Diagnosis: Hepatitis C virus infection

Treatment

The patient was considered for treatment with interferon 3 mU three times/week for 12 weeks.

Outcome of the treatment

- The patient responded well to the treatment, and his liver function parameters were normal
- He was advised for pre-marital counselling along with her fiancé to understand the burden of several diseases before and after marriage (also during or after pregnancy for females)

Take home message

- Hepatitis C virus infection is caused due to hepatitis C virus leading to inflammation of the liver
- Most of the patients with chronic hepatitis C infection are assymptomatic and may usually present fatigue or malaise
- Extrahepatic symptoms associated with hepatitis C virus infection include arthralgia, paraesthesias, myalgias, pruritus or sicca syndrome
- Six months of standard therapy with interferon (IFN) is considered to be excellent for treating patients with acute hepatitis C virus infection

CME – Post test

1. **Which of the following is not a typical symptom of secondary-stage syphilis?**
   a. Swollen glands
   b. Muscle and joint pain
   c. Dry eyes
   d. Rashes on the feet and hands
2. **When one parent has sickle cell trait and the other parent has two normal haemoglobin genes, what are the chances of the child being normal?**
   a. 10%
   b. 25%
   c. 50%
   d. 75%
3. **Which of these signs of severe anaemia can be seen during early childhood?**
   a. Jaundice
   b. Dark urine
   c. Protruding abdomen, with enlarged spleen and liver
   d. All of the above
4. **What proportion of people get rid of hepatitis C virus without treatment?**
   a. 50–70%
   b. 40–60%
   c. 15–25%
   d. 75%
5. **The secondary stage of syphilis typically starts with _____?**
   a. Development of a chancre
   b. Development of a rash
   c. Marked fatigue
   d. Severe abdominal pain
6. **What percentage of the infected individuals starts developing symptoms after 3–4 weeks of contracting HIV?**
   a. 30–40%
   b. 50–70%
   c. 70–90%
   d. 90–95%
7. **Which of the following are true of mother-foetus Rh incompatibility problems?**
   a. They can be prevented by injecting Rho-GAM into the mother's blood system
   b. They are much less likely to occur during the first pregnancy compared to later pregnancies
   c. Medical treatment can be nearly 100% effective in preventing such problems
   d. All of the above
8. **Which of the following is true of Rh+ people?**
   a. They are either homozygous dominant (DD) or heterozygous (Dd) for this trait
   b. They are all homozygous dominant (DD)
   c. They are all homozygous recessive (dd)
   d. They are either homozygous recessive (dd) or heterozygous (Dd) for this trait
9. **Mother-foetus incompatibility problems result from...**
   a. The mother's antibodies agglutinating the foetus' Rh positive red blood cells
   b. The foetus' antibodies agglutinating its own red blood cells
   c. The foetus' antibodies agglutinating its mother's red blood cells
   d. None of the above
10. **In patients with HIV, the antibodies will appear on a blood test within ____ months for 97% of infected people.**
    a. 1
    b. 2
    c. 3
    d. 4

Ans: 1-c; 2-c; 3-d; 4-c; 5-b; 6-b; 7-d; 8-a; 9-a; 10-c

Clinical challenges – Pre-marital counselling: A medical approach

Case 1 – A case of thalassaemia in a young child

Case presentation

A 15-month-old girl child was brought to the emergency department for complaints of pallor since 1 month and difficulty in breathing since past 4 days. She was treated with several medications by other doctor, but did not show any significant change. In subsequent visits, the child's blood was examined and severe anaemia was diagnosed.

Her parents reported that she had received blood transfusion 2 times in the previous 8 months. Her haemoglobin levels were seen to be normal, but she again started showing presenting complaints.

Medical history

- Initially, she was diagnosed and treated for severe anaemia
- She had received blood transfusions twice till date
- She was reported to be receiving blood transfusions regularly for her condition
- No surgical history, accidents or injury was reported
- Medication history mainly included blood transfusion, folic acid and topical skin ointment

Family and personal history

- No known medical history of any haematological disorder was reported in her parents or siblings
- No other family members reported any major illnesses

On examination

- Child was ill-looking, irritable and showed fear of strangers
- Height: 68.2 cm
- Weight: 6.4 kg
- Temperature: 97°F
- Pulse rate: 111 beats/min
- Respiratory rate (RR): 27 breaths/min
- Skin examination: Pallor; pustules on the neck and back of the scalp
- Forehead examination: Frontal and parietal bossing
- Respiratory, cardiovascular, musculoskeletal findings were unremarkable.

Other investigations

- Hb: 4.86 g/dL; severe anaemia
- Negative results for oedema, lymphadenopathy, cyanosis and mouth ulcers were reported
- Blood smear examination: Marked microcytosis and anisopoikilocytosis (Figure 1)

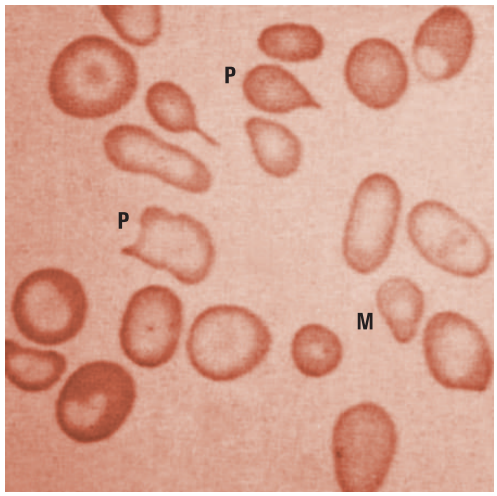

Figure 1: Blood smear showing marked microcytosis (M) and anisopoikilocytosis (P)

Diagnosis: Thalassaemia major with inadequate blood transfusion

Management and follow-up

- Recommended treatment for the child included lifelong regular blood transfusions to be administered every 2–5 weeks so as to maintain the pre-transfusion haemoglobin level above 9–10.5 g/dL
- Further, treatment with chelating agents were also recommended so as to prevent and treat haemosiderosis that may result due to repeated transfusions
- Regular follow-up and monitoring included assessment of CBC to check haemoglobin status, monitor vitals and growth and any complications
- Parents of the child were advised for regular check-ups, nutritional care, screening of future pregnancies, when to

receive chelation therapy, and importance of hepatitis B infection was also conveyed

Questions

1. **What is thalassaemia and how will you define its clinical and genetic classification?**
2. **How will you describe clinical picture of beta-thalassaemia?**

Case 2 – A case of congenital syphilis

Case presentation

An 8-month old male child was brought to the doctor's clinic by his parents for complaints of vomiting and diarrhoea since last 2 weeks. He was reported to be a full-term normal delivery.

Medical history

- The child did not report any major illness except the current complaining signs and symptoms
- He neither reported any allergy to any food product or any medication
- His immunisation schedule was reported to be up-to-date

Family history

- His mother presented history of syphilis during syphilis, which was treated with penicillin (antenatal card reported positive VDRL in 6th month of gestation)
- His father did not report any history of major illnesses
- No other family members reported any history of sexually transmitted diseases

On examination

- Malnourished and severely dehydrated
- Height: 51 cms
- Weight: 5.9 kg
- Temperature: 97.1°F
- RR: 26 breaths/min
- Head examination: Open fontanelle (5 cm x 5 cm)
- Skin examination: Peeling of skin of palms and soles (Figure 2)

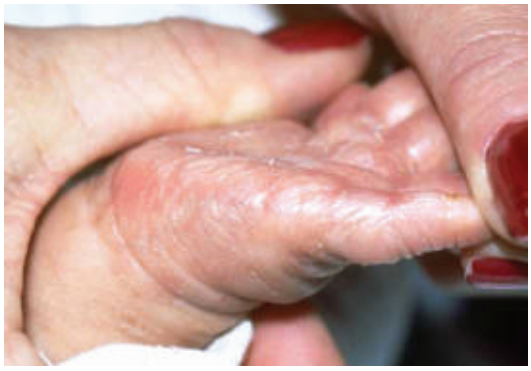

Figure 2: Lesions on skin of palm in congenital syphilis

- Respiratory, cardiovascular and musculoskeletal findings were unremarkable

Other investigations

- Hb: 11.1 g/dL
- Liver and renal function tests: Normal
- VDRL test: 1:1230; high
- CSF VDRL test: Negative
- HIV test: Negative
- Hepatitis C virus: Negative
- Chest X-ray: Normal

Diagnosis: Congenital syphilis

Treatment

The child was treated with Inj. Crystalline Penicillin for 21 days.

Outcome of the treatment

- On follow-up, VDRL test reported non-reactive results and his skin lesions had disappeared
- Parents were advised regular follow-ups

Questions

1. **What are the typical manifestations of congenital syphilis?**
2. **What are the complications of syphilis?**

Note: Answers to the clinical challenges will be given in the next issue.

Answers to clinical challenges – Management of common medical emergencies

Explanation to clinical challenges that have appeared in the issue dated June 01st–15th, 2014

Case 1 – A case of primary spontaneous pneumothorax

Questions

1. What are the risk factors for development of primary spontaneous pneumothorax?

Primary pneumothoraces arise in otherwise healthy people without any lung disease. Despite the absence of underlying pulmonary disease in patients with primary pneumothorax, subpleural blebs and bullae are found in up to 90% of cases at thoracoscopy, and in up to 80% of cases on CT scanning of the thorax. The aetiology of these bullous changes in otherwise apparently healthy lungs is unclear.

Smoking plays an important role and the lifetime risk of developing a pneumothorax in healthy smoking men may be 12% compared with 0.1% in non-smoking men. Also, tall thin individuals are more predisposed to the infection as alveoli at the lung apex in tall individuals are subject to significantly greater distending pressure than those at the base of the lung and are, therefore, more predisposed to the development of subpleural blebs.

2. What is the method of judging the size of a pneumothorax from chest radiograph?

The plain chest PA radiograph is a poor method of quantifying the size of a pneumothorax as it usually underestimates it. As per earlier guidelines, pneumothoraces were classified into three groups:

- "Small" defined as a "small rim of air around the lung";
- "Moderate" defined as lung "collapsed halfway towards the heart border"; and
- "Complete" defined as "airless lung, separate from the diaphragm

However, this method grossly underestimates the volume of the pneumothorax.

As per present guidelines, pneumothorax is regarded as small (< 2 cm) or large (≥ 2 cm) depending on the presence of a visible rim between the lung margin and the chest wall.

Case 2 – A case of acute ST elevation myocardial infarction

Questions

1. Why was primary PCI chosen over fibrinolysis in this case?

As per AHA guidelines, if immediately available, primary PCI should be performed in patients with STEMI (including true posterior MI) or MI with new or presumably new LBBB who can undergo PCI of the infarct artery < 12 hours of symptom onset, if balloon inflation occurred within < 90 mins of presentation, and if performed by experienced individuals at a center with cardiac surgery capability. Fibrinolysis is preferred if invasive strategy is not an option (cath lab delay/unavailability, vascular access difficulties, unavailability of skilled PCI lab) or if there are delays in an invasive strategy (prolonged transport, door-to-balloon time minus door-to-needle time is > 60 mins, or medical contact to balloon time is > 90 minutes). Fibrinolytic therapy should not be administered to patients with ST depression except when a true posterior (inferobasal) MI is suspected or when associated with ST elevation in lead aVR.

2. What are the indications of coronary artery bypass graft in MI patients?

As per guidelines of the American College of Cardiology (ACC) and the American Heart Association (AHA) the indications of CABG are as follows:

- Left main coronary artery stenosis > 50%
- Stenosis of proximal LAD and proximal circumflex > 70%
- 3-vessel disease in asymptomatic patients or those with mild or stable angina

- 3-vessel disease with proximal LAD stenosis in patients with poor left ventricular (LV) function
- 2-vessel disease and a large area of viable myocardium in high-risk area in patients with stable angina
- >70% proximal LAD stenosis with either ejection fraction < 50% or demonstrable ischemia on noninvasive testing

Other indications for CABG include the following:

- Disabling angina
- Ongoing ischemia in the setting of a non–ST segment elevation MI that is unresponsive to medical therapy
- Poor left ventricular function but with viable, nonfunctioning myocardium above the anatomic defect that can be revascularized

Medicolegal

ALLEGATIONS MADE IN THE COURT CANNOT BE DEFAMATION

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Litigants, during the course of litigation, can be very abusive both verbally and on paper. All kinds of defamatory remarks are put forth which are quite embarrassing. Matrimonial litigation is the worst. Allegations hurled can be absolutely defamatory but at the same time the litigants never take each other to the court for defamation for such atrocious allegations. This is because anything however defamatory pleaded before any judicial authority does not amount to defamation.

Legally speaking the litigants, their advocates, witnesses, juries and the judges have 'Absolute Privilege' to say whatever they want to say before a judicial authority.

Halsbury's Laws of England, Third Edition, volume 34, para no. 89 defines 'absolute privilege' thus:

Absolute Privilege: "No action lies, whether against judges, counsel, jury, witnesses, or parties, for words spoken in the ordinary course of any proceedings before any court or tribunal recognised by law. It manifests that the administration of justice would be paralysed if those who were engaged in it were liable to actions of libel or slander upon the imputation that they had acted maliciously and not bonafide. Thus, all witnesses or parties speaking with reference to the matter before the court have privilege for their evidence, whether oral or in writing, relevant or irrelevant, malicious or not. The privilege extends not only to words spoken but also to documents properly used and regularly prepared for use in the proceedings. Advocates are within the scope of this privilege, as also are juries and judges"

The Hon'ble Bombay High Court in a case Miss. Kamilini Manmade v. Union of India (1967) 69 BOM LR 512, has discussed the principle of absolute privilege in judicial proceeding. It holds that, "advocates, judges, witnesses and parties have absolute privilege for the words spoken during the course of judicial proceedings and that, claim for damages is not maintainable as the statements were made on an occasion which was absolute privilege. The rule of absolute privilege is based on sound public policy to enable a judge, party, witnesses, counsel or attorney to discharge his duty with a free mind uninfluenced by any fear of being sued for defamation"

In the above case, the appellant Miss. Kamilini Manmade had filed a suit against the Union of India and five others to recover Rs. 50,000/- as and by way of damages for defamation. A telephone directory was published at the instance of defendants no. l (Union of India) and no. 2, in which against the telephone number and the name of Miss Manmade the words "Miss Prostitution Solicitor" was printed in bold letters. This entry was prima facie defamatory. A suit was filed by Miss Manmade. The defendants apologised and pleaded that the entry appeared through inadvertence.

On one date, one of the defendants applied for an adjournment by making statement that, their clients have evidence in their possession about certain facts, i.e., the plaintiff (Miss Manmade) was a divorcee, she had been divorced by her husband on the ground of un-chastity and immorality and that, the plaintiff had a daughter, who had been married to one Bhavsav, but the plaintiff wanted her daughter to leave with some other person". These facts were stated to the Hon'ble Judge for getting the adjournment. Thereafter, the parties tired to settle the matter by amicable settlement. Miss Manmade was ready to accept an amount of Rs. 15,000/- which had been made on behalf of defendant no. 1 and 2.

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After few months, Miss. Manmade again approached the court for setting aside the settlement that had been arrived between the parties. The plaintiff, Miss. Manmade again filed the suit before the Hon'ble Bombay High Court against the Union of India and five other defendants claiming damages for the alleged defamation said to have been made by dependents no. 5 and 6 (the attorney and the counsel, who appeared for the defendant no. 1 and 2 in previous suit). The damages claimed for defamation was Rs. 1 lakh.

The Hon'ble Bombay High Court has quoted Halsbury's Laws of England, Third Edition Volume 34 para no. 277 which read as follows.

General Rule: No action will lie for defamatory statements, whether oral or written, made in the course of judicial proceedings before a court of justice or a tribunal exercising functions equivalent to those of an established court of justice. The authorities establish beyond all question this: that neither party, witness, counsel, jury nor judge, can be put to answer civilly or criminally for words spoken in office; that no action for libel or slander lies whether against judges, counsel, witnesses, or parties for words spoken in the course of any proceeding before any court recognised by law and this although the words were written or spoken maliciously, without any justification or excuse, and from personal ill will or anger against the party defamed. It is immaterial whether such proceedings take place in open court or in private, whether they are of a final or preliminary character, whether they are ex-parte or inter partes, and whether the court has jurisdiction to deal with the matter before it or not. The authorities are clear, uniform, and conclusive that, no action of libel or slander lies, whether against judges, counsel, witnesses, or parties, for words written or spoken in the ordinary course of any proceeding before any court or tribunal recognised by law.

Therefore, before every court established by law, the judge, advocates, witnesses, parties and attorneys are expected to execute their functions without fear. They are expected to act with free mind, uninfluenced by any fear of being sued for defamation. For this purpose the judicial system in India has incorporated the British principle of absolute privilege. If this principle is not incorporated, there will be chaos in our judicial system and in the courts of law as every litigation will give rise to a criminal prosecution for defamation.

Dr. Gopinath N. Shenoy is an Obstetrician and a Gynaecologist and a medicolegal consultant who exclusively defends the doctors in the Consumer Courts and the Medical Councils all over India. He was a Judge of the Consumer Court in Mumbai.

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